



NR0B1 gene

nuclear receptor subfamily 0 group B member 1

Normal Function

The *NR0B1* gene provides instructions for making a protein called DAX1. This protein plays an important role in the development and function of several hormone-producing (endocrine) tissues in the body. These tissues include the small glands located on top of each kidney (the adrenal glands), two hormone-secreting glands in the brain (the hypothalamus and pituitary), and the gonads (ovaries in females and testes in males). Before birth, the DAX1 protein helps regulate genes that direct the formation of these tissues. DAX1 also helps regulate hormone production in endocrine tissues after they have been formed.

Health Conditions Related to Genetic Changes

Swyer syndrome

X-linked adrenal hypoplasia congenita

More than 110 *NR0B1* mutations that cause X-linked adrenal hypoplasia congenita have been identified. Some of these genetic changes are deletions of all or part of the *NR0B1* gene. Other mutations lead to the production of an abnormally short version of the DAX1 protein. Still other mutations change single protein building blocks (amino acids) in a critical region of DAX1.

Most of the mutations responsible for X-linked adrenal hypoplasia congenita prevent the *NR0B1* gene from producing any active DAX1 protein. A shortage of DAX1 disrupts the normal development and function of hormone-producing tissues in the body. The main characteristics of this condition result when endocrine glands such as the adrenals, hypothalamus, pituitary, and gonads do not produce the right amounts of specific hormones.

other disorders

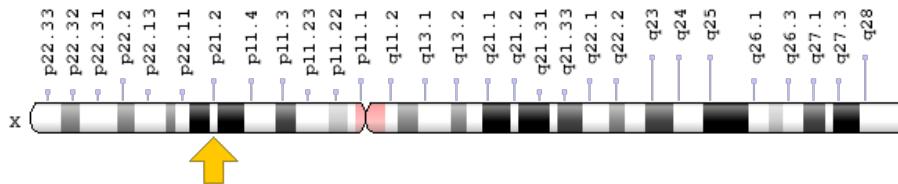
In some cases, genetic material is deleted from a region of the X chromosome that contains several genes, including *NR0B1*. This deletion results in a condition called adrenal hypoplasia congenita with complex glycerol kinase deficiency. In addition to the signs and symptoms of adrenal hypoplasia congenita, individuals with this condition may have delayed development and problems regulating their blood sugar levels. In rare cases, the deletion also includes the gene associated with Duchenne muscular dystrophy. People with this larger deletion have progressive

muscle weakness and wasting in addition to the other features of adrenal hypoplasia congenita with complex glycerol kinase deficiency.

Chromosomal Location

Cytogenetic Location: Xp21.2, which is the short (p) arm of the X chromosome at position 21.2

Molecular Location: base pairs 30,304,422 to 30,309,399 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AHC
- AHCH
- AHX
- DAX-1
- DAX1
- DSS
- gonadotropin deficiency
- GTD
- HHG
- NR0B1_HUMAN
- nuclear hormone receptor
- nuclear receptor DAX-1
- nuclear receptor subfamily 0, group B, member 1

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Phenotypic sex reversal in humans having two copies of the DAX1 locus
<https://www.ncbi.nlm.nih.gov/books/NBK9967/?rendertype=figure&id=A4121>

GeneReviews

- Nonsyndromic Disorders of Testicular Development
<https://www.ncbi.nlm.nih.gov/books/NBK1547>
- X-Linked Adrenal Hypoplasia Congenita
<https://www.ncbi.nlm.nih.gov/books/NBK1431>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28NR0B1%5BTIAB%5D%29+OR+%28DAX-1%5BTIAB%5D%29+OR+%28DAX1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- 46,XY SEX REVERSAL 2
<http://omim.org/entry/300018>
- GLYCEROL KINASE DEFICIENCY
<http://omim.org/entry/307030>
- NUCLEAR RECEPTOR SUBFAMILY 0, GROUP B, MEMBER 1
<http://omim.org/entry/300473>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/NR0B1ID44131chXp21.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NR0B1%5Bgene%5D>
- HGNC Gene Family: Nuclear hormone receptors
<http://www.genenames.org/cgi-bin/genefamilies/set/71>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7960

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/190>
- UniProt
<http://www.uniprot.org/uniprot/P51843>

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